Basics



What is it?

Points To Remember About Pachyonychia Congenita

- Pachyonychia congenita is very rare.
- It causes thick nails and painful calluses on the bottoms of the feet, and sometimes blisters on the palms of the hands.
- In about half of all cases, pachyonychia congenita is passed down from a parent. In the other cases, no one in the family has the disorder.
- There are no medicines for pachyonychia congenita, although over-the-counter drugs can help with pain.
- A primary care doctor or dermatologist (skin doctor) can help you manage symptoms.

Pachyonychia congenita is a very rare disorder. It causes thick nails and painful calluses on the bottoms of the feet. In some cases, blisters also form on the palms of the hands.

Who gets it?

Pachyonychia congenita is usually seen at birth or early in life. Anyone can have the disorder.

What are the types?

A change in any one of five genes can cause pachyonychia congenita. The type of disease gene mostly determines how severe the symptoms are.

What are the symptoms?

You may have symptoms for some time. Others may change, depending on your age, weight, activity level, and environment. Common symptoms of pachyonychia congenita include:

- Thick nails on the fingers and toes.
- Painful blisters and thick calluses on the soles of the feet when a child begins to walk. In some cases, blisters and calluses can form on the palms of the hands.

Other symptoms depend on the specific type of disorder and can include:

- Thick white patches on the tongue and inside the cheeks.
- Bumps around the hair follicles on the elbows, knees, and waistline.
- Fluid-filled lumps or cysts all over the body.
- Babies born with teeth that crumble and fall out soon after birth.

What causes it?

Pachyonychia congenita is caused by changes in at least one of five genes. These genes usually help make tough proteins in the skin, nails, and hair. The problem gene causes the nails to become thicker, and the skin blisters easily.

In about half of the cases, pachyonychia congenita is passed down from a parent. In the other cases, no one in the family has the disorder.

Is there a test?

To test for pachyonychia congenita, your doctor will take a sample of your saliva to look for one of the disease genes.

How is it treated?

There is no cure for pachyonychia congenita, and no medicines to treat it. Over-the-counter medicines are commonly used to treat painful symptoms.

Who treats it?

Because pachyonychia congenita is very rare, few doctors have experience treating it. Your best choices include:

- Dermatologist, who treats skin problems.
- Primary care doctor: Your own doctor knows your medical history, your lifestyle, and your special needs and can help as problems occur.

Living With It

There are no medicines for pachyonychia congenita. There are things you can do to cope with the disease. They include:

- Grinding or shaving down thick nails and skin. Take care that you don't make them too thin, as this could cause pain and infection.
- Wearing gloves to protect the hands while doing things like riding a bicycle or using hand tools.
- Wearing comfortable shoes and socks that keep your feet dry. This will help stop rubbing on painful calluses.

For More Info

U.S. Food and Drug Administration

Toll free: 888-INFO-FDA (888-463-6332)

Website: https://www.fda.gov

Drugs@FDA at https://www.accessdata.fda.gov/scripts/cder/daf. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics

Website: https://www.cdc.gov/nchs

NIH Office of Rare Diseases Research Genetic and Rare Diseases Information Center

Website: https://rarediseases.info.nih.gov

National Library of Medicine's Genetics Home Reference

Website: https://ghr.nlm.nih.gov/condition/

pachyonychia-congenita

Pachyonychia Congenita Project

Website: https://www.pachyonychia.org

American Academy of Dermatology

Website: https://www.aad.org

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- Asian Language Health Information
- Spanish Language Health Information

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<u>Understanding Pachyonychia Congenita</u> Ichthyosis Overview